

# Alpha Thalassemia (Hemoglobin Barts)

In September 2001, the State of Utah began screening for hemoglobinopathies. Hemoglobinopathy screening identifies infants with sickle cell anemia, as well as other hemoglobin disorders.

Hemoglobin Bart's is a relatively common hemoglobin variant detected by High Performance Liquid Chromatography (HPLC) testing and is only seen during the newborn period. Infants with an alpha-thalassemia syndrome usually show hemoglobin Bart's on the newborn screening test.

Alpha-thalassemia is characterized by a decrease in the rate of alpha chains production. Alpha chains are significantly important in normal hemoglobin formations, since they participate in the structure of all normal hemoglobins such A, A2, and F. Each is composed of two alpha and two non-alpha chains. This decrease in alpha chains production is caused by deletions of the alpha globin genes on chromosome 16. Normal individuals have four copies of the alpha-globin gene, 2 on each chromosome. **The loss of one to four of these genes is possible.** The presence of hemoglobin Bart's on the newborn screen almost always indicates that one or more of the baby's alpha-globin genes are deleted. The severity of the alpha-thalassemia condition is directly related to the amount of gene deletions present. See table.

Usual Genotypes	Alpha-Globin Gene Deletions		Clinical Features
$\alpha\alpha/\alpha\alpha$	0	Normal	
$-\alpha/\alpha\alpha$	1	Silent Carrier	This condition is clinically benign. If only one alpha gene is deleted, the other three genes can compensate nearly completely. These infants show normal CBC without microcytosis or anemia.
$--/\alpha\alpha$ or $-\alpha/-\alpha$	2	Alpha Thalassemia Trait	This condition, also referred to as alpha thalassemia minor, is characterized by microcytosis and hypochromia (low MCV, MCH) with <i>mild or no</i> anemia. The hypochromic anemia is often confused with iron deficiency anemia. Hemoglobin electrophoresis is usually normal and not helpful in the differential diagnosis. Iron supplementation is <b>not</b> recommended unless the patient has a diagnosis of iron deficiency anemia.
$--/-\alpha$	3	Hb H Disease	Three deleted alpha genes generally result in a moderately severe form of alpha-thalassemia. Clinical manifestations of this disorder are variable, but most patients are anemic and develop some degree of splenomegaly. These patients should be followed by a pediatric hematologist as they require frequent transfusions.
$--/--$	4	Fetal Hydrops	In fetal hydrops syndrome, also known as alpha thalassemia major, none of the alpha genes are present. This is not compatible with life. Death usually occurs in utero, unless intra-uterine blood transfusion is given, or during early infancy.

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